



TGFB1 gene

transforming growth factor beta 1

Normal Function

The *TGFB1* gene provides instructions for producing a protein called transforming growth factor beta-1 (TGF β -1). The TGF β -1 protein helps control the growth and division (proliferation) of cells, the process by which cells mature to carry out specific functions (differentiation), cell movement (motility), and the self-destruction of cells (apoptosis). The TGF β -1 protein is found throughout the body and plays a role in development before birth, the formation of blood vessels, the regulation of muscle tissue and body fat development, wound healing, and immune system function. TGF β -1 is particularly abundant in tissues that make up the skeleton, where it helps regulate bone growth, and in the intricate lattice that forms in the spaces between cells (the extracellular matrix). Within cells, this protein is turned off (inactive) until it receives a chemical signal to become active.

Health Conditions Related to Genetic Changes

Camurati-Engelmann disease

Approximately 10 mutations in the *TGFB1* gene have been found to cause Camurati-Engelmann disease. Most of the mutations change one protein building block (amino acid) in the TGF β -1 protein. The most common mutation replaces the amino acid arginine with the amino acid cysteine at position 218 in the TGF β -1 protein (written as Arg218Cys or R218C).

All mutations that cause Camurati-Engelmann disease result in a TGF β -1 protein that is always turned on (active). The overactive protein likely disrupts the regulation of bone growth and impairs muscle and body fat development. A disruption in the regulation of TGF β -1 activity can lead to increased bone density and other features of Camurati-Engelmann disease.

cancers

Some *TGFB1* gene mutations are acquired during a person's lifetime and are present only in certain cells. These changes are called somatic mutations and are not inherited. Somatic mutations in the *TGFB1* gene that cause alterations in the activity (expression) of the TGF β -1 protein are associated with certain cancers. The altered protein expression may enhance several cancer-related events such as

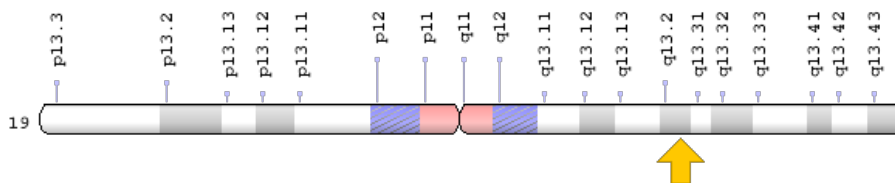
cell division (proliferation), cell motility, and the development of new blood vessels (angiogenesis) that nourish a growing tumor. The TGF β -1 protein is abnormally active (overexpressed) in certain types of prostate cancers. Altered TGF β -1 expression has also been found in breast, colon, lung, and bladder cancers.

A variation (polymorphism) in the *TGFB1* gene that changes a single amino acid in the TGF β -1 protein is associated with prostate cancer. In people with this polymorphism, the amino acid leucine is replaced with the amino acid proline at position 10 in the TGF β -1 protein. Although it has no apparent effect in healthy people or those with a condition caused by a different mutation in the *TGFB1* gene, this polymorphism is associated with accelerated disease progression and a poorer outcome in patients with prostate cancer.

Chromosomal Location

Cytogenetic Location: 19q13.2, which is the long (q) arm of chromosome 19 at position 13.2

Molecular Location: base pairs 41,330,531 to 41,353,933 on chromosome 19 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CED
- diaphyseal dysplasia 1, progressive
- DPD1
- TGF-beta-1
- TGF-beta 1 protein
- TGFB
- TGFB1_HUMAN
- TGFbeta

- transforming growth factor-beta 1
- transforming growth factor, beta 1
- transforming growth factor, beta 1 (Camurati-Engelmann disease)

Additional Information & Resources

Educational Resources

- Cancer Medicine (sixth edition, 2003): Transforming Growth Factor- β (TGF- β)
<https://www.ncbi.nlm.nih.gov/books/NBK12565/#A4309>
- Eureka Bioscience Collection: TGF β Signaling
<https://www.ncbi.nlm.nih.gov/books/NBK6525/#A31193>
- Molecular Cell Biology (fourth edition, 2000): TGF β signaling pathway
<https://www.ncbi.nlm.nih.gov/books/NBK21715/?rendertype=figure&id=A6758>

GeneReviews

- Camurati-Engelmann Disease
<https://www.ncbi.nlm.nih.gov/books/NBK1156>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28TGFB1%5BTIAB%5D%29+OR+%28TGF+beta-1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- TRANSFORMING GROWTH FACTOR, BETA-1
<http://omim.org/entry/190180>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
<http://atlasgeneticsoncology.org/Genes/TGFB1ID42534ch19q13.html>
- Cancer Genetics Web
<http://www.cancerindex.org/geneweb/TGFB1.htm>
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=TGFB1%5Bgene%5D>
- HGNC Gene Family: Endogenous ligands
<http://www.genenames.org/cgi-bin/genefamilies/set/542>

- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=11766
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/7040>
- UniProt
<http://www.uniprot.org/uniprot/P01137>

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